

Author Index to Volume 47

(A) Allen Award address; (ASHG) = American Society of Human Genetics report; (BR) = Book Review; (E) = Editorial; (HGES) = Human Genetics Education section; (L) = Letter to the Editor

- Ackford, H., 536
 Adelsberger, P. A., 968
 Al-Ali, A., 1013
 Allen, L., 236, 828
 Alpert, E., 583 (L)
 Amati, P., 904
 Amos, C. I., 247, 842
 Andermann, E., 815
 Anderson, L. L., 952
 Angelico, F., 429
 Anglani, F., 169 (L)
 Ankra-Badu, G., 1013
 Annerén, G., 595 (BR)
 Antonarakis, S. E., 968
 Antonelli, A., 228
 Antonini, R., 429
 Apold, J., 1002
 Arai, Y., 562
 Archer, B. T., III, 551
 Arets, A., 196
 Arinami, T., 988
 Ashton, L. J., 802
 Azen, E. A., 686

 Bähr-Porsch, S., 656
 Baker, E., 187, 493, 802
 Bakken, A., 1002
 Bale, A. E., 389
 Ballabio, A., 664
 Barbujani, G., 867
 Barr, M., Jr., 745 (BR)
 Barsh, G., 596 (BR)
 Bartels, I., 656
 Basdevant, A., 721
 Bashir, R., 536, 941
 Baumiller, R. C., 763 (HGES-BR)
 Bayleran, J., 815
 Bean, B., 351 (L)
 Beaudet, A. L., 603 (E), 611
 Bell, M. V., 181
 Berenson, G. S., 247
 Berrebi, A., 1013
 Berson, E. L., 790
 Best, S., 369
 Beutler, E., 575, 1008

 Bhattacharya, S. S., 536, 935, 941
 Bick, D. P., 740 (L)
 Bingham, E. L., 616
 Binkert, F., 968
 Bird, A. C., 536
 Bird, C. C., 982
 Bird, T. D., 915
 Bishop, D. F., 784
 Black, S. H., 740 (L)
 Blake, E., 515
 Blangero, J., 414
 Bloch, M., 4
 Bodkin, M., 440
 Boehnke, M., 218, 470, 616, 1031 (BR)
 Bogart, M. H., 353 (L)
 Bohlman, M. C., 1023 (L)
 Boman, H., 1002
 Bond, A., 13
 Bonney, G. E., 247, 542
 Bordoni, A., 904
 Borecki, I. B., 542
 Borgmann, S., 656
 Botstein, D., 887 (A)
 Bottema, C. D. K., 202, 835
 Boucher, M. C., 611
 Bouhassira, E. E., 161
 Boulay, B., 815
 Brackmann, H. H., 743 (L)
 Bradley, D. G., 941
 Bresolin, N., 904
 Bridge, P. J., 202
 Brock, D., 164 (L)
 Brooks, D. A., 802
 Brooks-Wilson, A. R., 952
 Brown, A., 454
 Brown, B. I., 735
 Brown, G. K., 286
 Brown, R. M., 286
 Brown, W. T., 175 (E)
 Brunsman, F., 622
 Buchris, V., 20
 Buetow, K., 13
 Buxbaum, J. M., 127

 Callen, D. F., 187, 493, 802
 Camporese, C., 169 (L)
 Capua, A., 815
 Carothers, A. D., 165 (L)
 Carson, N. L., 946
 Cerino, A., 228
 Chakraborty, R., 87
 Chakraborty, S., 37
 Chakravarti, A., 635, 644, 926
 Chamberlain, J. S., 795
 Chance, P. F., 915
 Chapman, M., 1 (E)
 Charmley, P., 860
 Chase, C. L., 266
 Chen, S.-H., 1020
 Chen, Y.-T., 735
 Chimera, J., 515
 Christianson, R. E., 727
 Civelli, O., 828
 Claussen, U., 181
 Clow, C., 815
 Cole, D. E. C., 776
 Coleman, M., 935
 Concannon, P., 45, 860
 Constantinou, C. D., 670
 Cophignon, J., 877 (L)
 Cotton, R. G. H., 279
 Couturier, J., 877 (L)
 Cox, T. M., 101
 Craig, I., 935
 Cremers, F. P. M., 20, 622
 Cross, N. C. P., 101
 Crystal, R. G., 403
 Cunningham, G., 583 (L)
 Cunningham, G. C., 899
 Cystic Fibrosis Genetic Analysis Consortium, The, 354 (L)

 Dahl, H.-H. M., 279, 286
 Dahl, N., 187, 275
 Daigneault, J., 606
 Dalla Piccola, B., 664
 D'Amico, C., 376
 Davies, K., 935
 Davies, K. E., 181

- Davis, J., 896 (E)
 Dawson, D. V., 842
 De Braekeleer, M., 302, 580
 (L), 606, 815
 De Jonghe, P., 680
 Deka, R., 644
 de la Chapelle, A., 622
 Delattre, O., 877 (L)
 Del Ben, M., 429
 Del Junco, D., 583 (L)
 Denko, N., 459
 Derome, P., 877 (L)
 Desnick, R. J., 784
 De Winter, G., 680
 Diala, E. S., 376
 Dickens, B., 4
 Di Donato, S., 228, 904
 DiMartino, N. A., 854
 Ding, J.-H., 735
 Di Silvestre, D., 706
 Dones, I., 228
 Donnai, D., 166 (L)
 Dorsey, B. V., 376
 Duband, J. L., 308
 Ducat, L., 1029 (L)
 Duffy, D. L., 590 (L)
 Dunlop, M. G., 982
 Dyer, T. D., 414

 Edwards, J. H., 1024 (L)
 Egli, H., 743 (L)
 Eiben, B., 656
 Eiken, H. G., 1002
 Eisenbarth, G., 1029 (L)
 Elston, R. C., 247, 842
 Emi, M., 107, 721
 Epstein, C. J., 236, 601 (E)
 Erikson, A., 275
 Erlich, H. A., 515
 Erway, L. G., 760 (HGES-BR)
 Esakowitz, L., 536
 Evans, H. J., 982
 Eyre, H. J., 493, 802

 Falls, K., 376
 Farrar, G. J., 941
 Fedde, K. N., 767, 776
 Fefelova, V. V., 294
 Ferrell, R. E., 635, 644
 Ferretti, L., 228
 Fildes, N., 515
 Fischel-Ghodsian, N., 1023 (L)

 Foellmer, B. E., 483
 Folstein, S. E., 362 (BR)
 Fontaine, B., 823
 Foroud, T., 860
 Forrest, S. M., 279
 Fox, E. A., 973
 Francke, U., 483, 551
 Franco, B., 926
 Fredriksen, Å., 1002
 Freemantle, C. J., 493
 Frias, J. L., 167 (L)
 Fucharoen, S., 369
 Fujita, R., 228
 Fujiwara, T. M., 606
 Fukuhara, Y., 784

 Gadbois, P., 329
 Gagné, R., 329, 815
 Gail, M. H., 499
 Gal, A., 941
 Gambert, P., 721
 Garcia, C., 926
 Garel, L., 338
 Garver, K. L., 345 (ASHG)
 Gatti, R. A., 860
 Gatz, G., 656
 Geier, M. R., 740 (L)
 Geisterfer-Lowrance, A. A. T.,
 389
 Gellera, C., 904
 Gellert, G., 656
 Gheuens, J., 680
 Giaccia, A. J., 459
 Gieselmann, V., 880 (L)
 Gilbert, D. A., 499
 Ginsburg, D., 616
 Glatt, K., 149
 Glorieux, F. X., 28
 Goebel, R., 656
 Goldgar, D. E., 957
 Golla, A., 664
 Gonzalez, F. J., 994
 Goodfellow, P. J., 952
 Goodman, H. O., 446, 454
 Gorevic, P. D., 127
 Gorodezky, C., 515
 Grabowski, G. A., 79
 Graham, J. M., Jr., 149
 Grandy, D. K., 828
 Graveline, R., 376
 Greenberg, F., 583 (L)
 Grenier, A., 325, 329

 Groffen, J., 706
 Gusella, J. F., 790, 823

 Haan, E. A., 493
 Hackel, E., 762 (HGES-BR)
 Hagerman, P. J., 876 (L)
 Halket, J., 329
 Hammans, W., 656
 Hanauer, A., 228
 Hanioka, N., 994
 Hansen, L. L., 286
 Hansmann, I., 656
 Hanson, M. P., 823
 Harris, R., 750 (HGES-L)
 Hart, I., 459
 Haseltine, F. P., 364 (BR)
 Hata, A., 107, 721
 Hauselman, E., 644
 Hay, R. J., 499
 Hayden, M., 4
 Hecht, B. K., 745 (BR)
 Hecht, F., 745 (BR)
 Hechtman, P., 815
 Hedrick, A., 4
 Hegele, R., 107
 Helmuth, R., 515
 Hentemann, M., 656
 Herdman, R. C., 1028 (L)
 Herndon, C. N., 446
 Hesketh, C., 369
 Hett, G., 524
 Heye, T., 664
 Higuchi, R., 515
 Hildesheimer, M., 20
 Hiller, G., 389
 Hilman, B. C., 606
 Hirsch, P. C., 1023 (L)
 Hirschhorn, R., 73, 440
 Hirst, M. C., 181
 Hoffner, L., 635
 Hogan, C., 376
 Holden, J. J. A., 20, 395
 Hook, E. B., 581 (L), 741 (L)
 Hopwood, J. J., 187, 802
 Hori, K., 562
 Horn, G. T., 606
 Horsthemke, B., 181
 Howell, N., 629
 Howells, D. W., 279
 Hsieh, C.-L., 483
 Huether, C. A., 748 (HGES-E)
 Huff, V., 155

- Huggins, M., 4
 Humphries, M. M., 941
 Humphries, P., 941
 Humphries, S. E., 429
 Hyland, V. J., 187

 Iida, R., 121
 Ikawa, K., 236
 Ikehara, Y., 121
 Ikeuchi, T., 236
 Inglehearn, C. F., 536, 941
 Iverius, P.-H., 107, 721

 Jackson, C. E., 946
 Jacobs, K., 680
 Jacobsen, S. J., 376
 Jacobson, D. R., 127
 Jacquez, G. M., 867
 Jansen, R., 808
 Jarcho, J. A., 389
 Jay, B., 622
 Jay, M., 536, 622, 935, 941
 Jewell, A. F., 823
 Johnston, P. A., 551
 Jones, O. W., 353 (L)

 Kaback, M. M., 698
 Kajihara, S., 562
 Kajii, T., 988
 Kammerer, C. M., 414
 Kanani, S., 4
 Kaplan, F., 815
 Kaplan, L., 149
 Kappler, J., 880 (L)
 Kasperczyk, A., 854
 Kaufman, D. L., 790
 Kawashima, H., 236
 Keats, B. J. B., 247
 Kenna, P., 941
 Kerem, B.-s., 606
 Ketterling, R. P., 202, 835
 Khoury, M. J., 742 (L)
 Kidd, K. K., 946
 Kim, H.-S., 686
 Kimura, S., 994
 Kishi, K., 121
 Kitagawa, T., 562
 Kizer, K. W., 899
 Klinger, K., 606
 Klitz, W., 515
 Knoll, J. H. M., 149
 Knowles, M. R., 611

 Knowlton, R. G., 37
 Kobayashi, K., 611
 Koch, R., 706
 Kochhan, L., 743 (L)
 Koeberl, D. D., 202
 Kolodny, E., 815, 881 (L)
 Kontusaari, S., 112
 Korenberg, J. R., 236
 Krontiris, T. G., 854
 Kuhl, W., 575, 1008
 Kuivaniemi, H., 112
 Kuo, C.-Y., 616
 Kupper, L. L., 266
 Kurdi-Haidar, B., 1013
 Kuwano, A., 988
 Kwiatkowski, D. J., 62

 Laberge, C., 308, 325, 329
 Labuda, M., 28
 Ladda, R. L., 112
 Lafreniere, R. G., 551
 Lagerström, M., 275
 Lalande, M., 149
 Lalouel, J.-M., 107, 721, 915
 Lambert, M., 815
 Lange, K., 860
 Larochelle, J., 302, 338
 Latham, T., 79
 Lathrop, G. M., 542
 Latreille, P., 686
 Lebo, R. V., 583 (L)
 Ledley, F. D., 808
 Lehner, T., 20
 Lenaerts, C., 338
 Lent, K. M., 345 (ASHG)
 Leone, M., 228
 Leppert, M., 915
 Lernmark, Å., 1029 (L)
 Lescault, A., 308, 329
 Lester, D. H., 536
 Leto, T. L., 483
 Letter, F., 308
 Levilliers, J., 664
 Lewis, R. A., 13
 Ligi, L., 867
 Lillicrap, D. P., 202
 Lim, J., 698
 Lindsay, S., 935
 Lipe, H., 915
 Litt, M., 828, 935
 Litt, R., 828
 Litvak, G., 20

 Lomax, K. J., 483
 Lorens, J. B., 1002
 Luc, G., 721
 Luce, M. C., 515
 Ludecke, H. J., 181
 Ludwig, E. H., 712
 Ludwig, M., 742 (L), 941
 Lupski, J. R., 926
 Lustig, L., 581 (L)
 Luzzatto, L., 1013
 Lynch, A., 4

 McAfee, M., 62
 McBride, W. O., 389
 McCabe, E. R. B., 795
 McCarthy, B. J., 712
 McClatchey, A. I., 790
 MacCluer, J. W., 414
 McCullough, D., 629
 McFadden, R. R., 171 (BR)
 McGill, H. C., Jr., 414
 McGuffin, P., 524
 McKeag, D. B., 753 (HGES)
 McKenna, W., 389
 MacKinnon, R. N., 181
 MacLaren, R., 459
 McLean, W. T., Jr., 446
 McWilliam, P., 941
 Maddelena, A., 740 (L)
 Madej, R., 515
 Magenis, E., 236
 Magenis, R. E., 149, 828
 Majumder, P. P., 644
 Malamut, R. I., 926
 Malech, H. L., 483
 Malhotra, U., 860
 Mandel, J.-L., 228
 Maragos, C., 286
 Marchbanks, R. M., 524
 Martin, G. M., 364 (BR)
 Martin, J. J., 680
 Martin, N. G., 137, 590 (L)
 Martin, R. H., 349 (L)
 Martiniuk, F., 73, 440
 Martuza, R. L., 823
 Mason, P. J., 1013
 Mathews, J. D., 590 (L)
 Mazzarella, B., 429
 Meadows, A., 155
 Mehler, M., 73
 Meitinger, T., 664
 Mélançon, M. J., 580 (L)

- Melançon, S., 815
 Meredith, G., 73
 Meyer, U. A., 994
 Miciak, A., 1024 (L)
 Miller, N., 429
 Minaguchi, K., 686
 Mirman, D., 459
 Mitchell, A. L., 389
 Moggio, M., 904
 Montes de Oca-Luna, R., 926
 Moraine, C., 664
 Morgan, K., 28, 606
 Morissette, J., 325, 329
 Morris, C. P., 187, 802
 Morton, C. C., 389, 973
 Mott, G. E., 414
 Mukai, T., 562
 Mulinare, J., 742 (L)
 Mulligan, L. M., 395
 Murken, J., 664
 Myerowitz, R., 169 (L)
 Myers, S., 952

 Nagao, Y., 784
 Nagel, R. L., 161
 Nakamura, Y., 982
 Nanjee, M. N., 429
 Navon, R., 881 (L)
 Neufeld, E. F., 698
 Nicholls, R. D., 149
 Nikoskelainen, E. K., 95
 Nussbaum, R., 13

 Oberle, I., 187
 O'Brien, S. J., 499
 O'Brien, W. E., 611
 O'Connell, P., 915
 Odland, E., 1002
 Oechsli, F. W., 727
 Ogasawara, N., 236
 Ohashi, H., 988
 Oldenburg, J., 743 (L)
 Olek, K., 743 (10)
 Olson, J. M., 470
 Oostra, B. A., 187
 Oppenheim, A., 1013
 O'Regan, S., 317
 Oshima, A., 784
 Osmers, R., 656
 Ott, J., 20, 166 (L), 1029 (L)
 Owada, M., 562
 Özçelik, T., 551

 Pack, M., 670
 Pancoast, I., 454
 Pandolfo, M., 228
 Pandya, A., 706
 Pannacci, M., 904
 Papiha, S. S., 536
 Paradis, K., 338
 Patel, P. I., 926
 Paw, B. H., 698
 Pawlowitzki, I. H., 622
 Pee, D., 499
 Permutt, M. A., 1029 (L)
 Petersen, M. B., 968
 Petit, C., 664
 Pettersson, U., 275
 Philippon, J., 877 (L)
 Picci, L., 169 (L)
 Pillers, D.-A. M., 795
 Pinckers, A. J. L. G., 622
 Plante, M., 308
 Poon, R., 395
 Potier, M., 815
 Powell, B. R., 795
 Prentice, R., 255
 Prevost, C., 815
 Prockop, D. J., 112, 670
 Proia, R. L., 881 (L)
 Propping, P., 880 (L)
 Pujades, M. A., 575
 Pulst, S.-M., 236
 Punnett, H. H., 568

 Quarrell, O. W. J., 4

 Raeymaekers, P., 680
 Rainer, J., 795
 Ramesh, V., 790
 Rao, D. C., 542
 Rasmussen, S., 742 (L)
 Rauskolb, R., 656
 Read, A. P., 166 (L)
 Rediker, K., 376
 Redmond, R., 941
 Reefer, J., 644
 Reid, Y. A., 499
 Riccardi, V. M., 155
 Ringenbergs, M. L., 493
 Rising, M. B., 376
 Robertson, M., 107
 Robinson, A., 363 (BR)
 Rocchi, M., 187
 Romain, D., 493

 Ropers, H.-H., 187, 622
 Roses, A. D., 596 (BR)
 Roth, M. S., 616
 Rouleau, G. A., 823, 877 (L)
 Roy, C. C., 338
 Rozen, R., 606
 Rubinstein, P., 1029 (L)
 Russo, P., 317

 Sakuraba, H., 784
 Salser, W., 860
 Samanns, C., 941
 Sanal, O., 860
 Sandkuyl, L., 20
 Sankila, E.-M., 622
 Sanson, M., 877 (L)
 Sato, W., 121
 Saunders, G. F., 155
 Savill, J., 429
 Savontaus, M.-L., 95
 Sawazaki, K., 121
 Scarlato, G., 904
 Schinzel, A., 941, 968
 Schmidt, M., 187
 Schmill, N., 515
 Schonberg, S. A., 236
 Schulman, J. D., 740 (L)
 Schultz, P., 376
 Schwaab, R., 743 (L)
 Schwartz, C. E., 187, 446, 454
 Schwartz, M., 622
 Schwartz, R. H., 606
 Scott, C. R., 1020
 Scott, H. S., 802
 Scliver, C., 815
 Seashore, M. R., 759 (HGES-BR)
 Seidman, C. E., 389
 Seidman, E. G., 338
 Seidman, J. G., 389
 Seizinger, B. R., 823
 Senger, G., 181
 Sergeant, M., 524
 Servidei, S., 904
 Shapiro, L. J., 583 (L)
 Sharp, E. M., 941
 Shih, V. E., 790
 Shiloh, Y., 20
 Shimmoto, M., 784
 Shore, S., 169 (L)
 Sieving, P. A., 616
 Simensen, R. J., 446
 Simons, M. J., 583 (L)

- Simpson, N. E., 946, 952
 Sippell, W. G., 664
 Sirugo, G., 228
 Slaugenhaupt, S., 926
 Smeets, D., 196
 Smith, F. I., 79
 Smouse, P. E., 761 (HGES-BR)
 Social Issues Committee, ASHG,
 343 (ASHG)
 Solomon, S. D., 389
 Sommer, S. S., 202, 835
 Sood, R., 395
 Spielman, R. S., 45, 1029 (L)
 Stamato, T. D., 459
 Stambolian, D., 13
 Stanislovitis, P., 606
 Steel, C. M., 982
 Stevenson, R. E., 446, 454
 Stoneking, M., 515
 Strong, L. C., 155
 Südhof, T. C., 551
 Surti, U., 635, 644
 Sutherland, G. R., 187, 802
 Suthers, G. K., 53, 187
 Suzuki, K., 568
 Suzuki, Y., 784
 Swerts, L., 680
 Swift, M., 266
 Swinford, A. E., 753 (HGES)
 Sykes, B., 593 (L)
 Sylvester, D. R., 45
 Szabo, P., 20

 Takahashi, H., 403
 Talmud, P. J., 429
 Tanaka, A., 568
 Tanguay, R. M., 308
 Taniguchi, N., 236
 ten Kate, L. P., 359 (L)
 Theilmann, J., 4

 Thein, S. L., 369
 Theophilus, B. D. M., 79
 Thomas, G., 877 (L)
 Thomas, I. T., 167 (L)
 Thomas, N. S., 187
 Thompson, E., 255
 Tidmarsh, S., 524
 Tieu, P. T., 698
 Timmerman, V., 680
 Tobin, A. J., 790
 Torfs, C. P., 727
 Towbin, J. A., 795
 Treloar, S. A., 137
 Tromp, G., 112
 Tsui, L.-C., 606
 Tsukahara, M., 988
 Tzall, S., 73, 440

 Ulmer, J., 454

 Valet, J. P., 308, 325
 Vallee, B. L., 973
 Van Broeckhoven, C., 680
 van den Berg, B. J., 727
 Vandenberghe, A., 680
 van de Pol, D. J. R., 622
 Villki, J., 95
 Vives-Corrons, J.-L., 575
 Vosberg, H.-P., 389

 Waldren, C., 459
 Waller, K., 581 (L)
 Wasi, P., 369
 Watson, J. E. V., 181
 Watty, A., 941
 Waurin, J. L., 353 (L)
 Weatherall, D. J., 369
 Weber, A., 338
 Weeks, D. E., 166 (L), 592 (L)
 Wei, S., 860

 Weiffenbach, B., 376
 Weitnauer, L., 228
 Wenger, D. A., 37
 Weremowicz, S., 973
 West, R., 236
 Whatley, S., 524
 White, B. N., 20, 395
 White, C., 499
 White, R. L., 892 (A), 982
 Whyte, M. P., 767, 776
 Weiringa, B., 622
 Willard, H. F., 551
 Wilson, P. J., 187
 Wilson, S. R., 53
 Winichagoon, P., 369
 Wright, A. F., 536, 622, 935
 Wright, J. A., 45
 Wright, L. G., 45
 Wu, D., 795
 Wu, J., 946
 Wyllie, A. H., 982

 Xu, C.-F., 429

 Yamamoto, K., 236
 Yang, B.-Z., 735
 Yaouanq, J., 542
 Yasuda, T., 121
 Yoon, H.-S., 835
 Young, J. L., 740 (L)
 Young, M. R., 616
 Young, S. B., 670
 Youssoufian, H., 62

 Zacchello, F., 169 (L)
 Zeviani, M., 904
 Zhao, L. P., 255
 Zhou, Q.-Y., 828
 Ziv, Y., 20
 Zlotogora, J., 37

Subject Index to Volume 47

(E) = Editorial; (HGES) = Human Genetics Education section; (L) = Letter to the Editor

- Abortion, spontaneous, cytogenetic analysis, chorionic villi, 656
- Acid alpha glucosidase
 - Allele GAA 2, glycogen and transient gene expression, 440
 - Deficiency, heterogeneity, 73
- Acid β -glucosidase gene, alleles in Gaucher disease, 79
- Adenomatous polyposis, familial, polymorphic DNA markers, 982
- Affection status, segregation/linkage analysis, genetic hemochromatosis, 542
- Åland Island eye disease, deletion mapping, p21, 795
- Albinism-deafness syndrome, X-linked, mapping, 20
- Aldolase B gene
 - Nonsense mutation, hereditary fructose intolerance, 562
 - Partial deletions in hereditary fructose intolerance, 101
- Alkaline phosphatase
 - Cultured skin fibroblasts, 776
 - Normal and hypophosphatasia fibroblasts, 767
- Allan-Herndon syndrome
 - Clinical studies, 446
 - Xq21 DNA markers, linkage, 454
- Allele(s)
 - Acid alpha glucosidase allele, base-pair substitution, 440
 - Acid β -glucosidase gene in Gaucher disease, 79
 - α 1-Antitrypsin deficiency, deletion of α 1-antitrypsin coding exons, 403
 - Chromosome 22, meningioma, 877 (L)
 - Hex A mutant, frequency, Tay-Sachs screening, 698
 - Minisatellite, diversification, HRAS1 locus, 854
 - Mutant
 - CYP2D6, defective oxidation of drugs, 994
 - Hex A, frequency, Tay-Sachs screening, 698
 - Tay-Sachs disease, 815
 - PRB3 locus, proline-rich gl 8, 686
- Allele frequency, HLA-DQ α , 515
- Allele-specific PCR, hemoglobin C mutation, 1023 (L)
- Allen Award addresses 1989, 887, 892
- α -Actinin gene, linkage to β -spectrin gene, 62
- α/β Locus, T cell receptor, 14q11, 973
- α -Galactosidase A gene, point mutations, Fabry disease, 784
- α -L-Iduronidase gene, 4p16.3, 802
- α Subunit gene, β -hexosaminidase, point mutation, infantile Tay-Sachs disease, 568
- α 1-Antitrypsin
 - Deficiency allele, deletion of α 1-antitrypsin coding exons, 403
 - Nullisoma di prociada, α 1-antitrypsin coding exons, 403
- α 1-Glycine, substitution by cysteine in COL1A1, osteogenesis imperfecta, 670
- Alu-PCR, chromosome 17-retaining hybrids, Charcot-Marie-Tooth disease type 1A gene marker, 926
- American Society of Human Genetics
 - Allen Award addresses, 887, 892
 - Membership survey results, 345
 - MSAFP policy statement update, remarks on criticism, 740 (L)
 - Social Issues Committee report, 343
- Amyloidosis, senile systemic, homozygous, transthyretin variant, 127
- Aneurysms, familial, inheritance of RNA splicing mutation in COL3A1, 112
- Angelman syndrome, DNA markers, 149
- Angiogenin gene, 14q11, 973
- Anosmia, Xp22.3, 664
- Antibodies, thyroid, Down syndrome and other trisomies, 727
- Antibody method, BrdUrd, replication patterns of fragile X, 988
- Antigen, HLA, distribution, ethnogeny, 286
- Apolipoprotein(s)
 - A-I, dietary environment, 414
 - AI-CIII-AIV gene cluster, lipoprotein and apolipoprotein levels, 429
- B
 - Gene loci, lipoprotein and apolipoprotein levels, 429
 - Mutation, haplotype analysis, 712
 - B100, familial defective, apo B mutation, 712
- Appendectomy, twins, 590 (L)
- Ataxia-telangiectasia locus, mapping, 11q23, 860
- Base-pair substitution, acid alpha glucosidase allele, 440
- β^s Chromosomes, Bantu haplotype, deletion, 161
- β -Hexosaminidase α subunit gene, point mutation, infantile Tay-Sachs disease, 568
- β -spectrin gene, linkage to α -actinin gene, 62
- β -Thalassemia, prenatal diagnosis, 369
- Birth defects, 741 (L), 742 (L)
- Books reviewed, author(s)/editor(s):
 - Boyd, G. W., 745

- Burch, J. E., 364
 Burnham, D., 763 (HGES)
 Chesters, M. S., 364
 Clark, A. G., 761 (HGES)
 Clegg, M. T., 762 (HGES)
 Comings, D. E., 362, 363
 Elston, R. C., 1031
 Fodden, M., 763 (HGES)
 Fristrom, J. W., 762 (HGES)
 Gilbert, S. G., 760 (HGES)
 Gordon, S., 763 (HGES)
 Grumbach, M. M., 595
 Harper, P. S., 596
 Hartl, D. E., 761 (HGES)
 Hodge, S. E., 1031
 Hubbard, R., 364, 763 (HGES)
 Hughley, B., 763 (HGES)
 Lowe, M., 763 (HGES)
 Lyon, M. F., 596
 MacCluer, J. W., 1031
 Mange, A. P. and E. J., 759 (HGES)
 Office of Technology Assessment, Congress of the United States, 171
 Persaud, P. V. N., 745
 Rosenfeld, R. G., 595
 Rosoff, B., 763 (HGES)
 Russell, P. J., 762 (HGES)
 Searle, A. G., 596
 Spence, M. A., 1031
 Tobach, E., 763 (HGES)
 Vroman, G., 763 (HGES)
- Books reviewed, title:
Biological Basis of Disease: Selected Papers by P. R. J. Burch, The, 364
Environmental Causes of Human Birth Defects, 745
Genes and Gender I, 763 (HGES)
Genes and Gender II: Pitfalls in Research on Sex and Gender, 763 (HGES)
Genes and Gender III: Genetic Determinism and Children, 763 (HGES)
Genes and Gender IV: The Second X and Women's Health, 763 (HGES)
Genes and Gender V: Socialization toward Inequity, 763 (HGES)
Genes and Gender VI: The Gendered Face of Peace and War: A Challenge to Genetic Determinism, 763 (HGES)
Genetic Strains and Variants of the Laboratory Mouse, 2d ed., 596
Genetics, 2d ed., 762 (HGES)
Genetics: Human Aspects, 759 (HGES)
- Myotonic Dystrophy*, 2d ed., 596
New Developments in Biotechnology, 171
On Stress, Disease and Evolution, 745
Pictorial Human Embryology, 760 (HGES)
Politics of Women's Biology, The, 364
Principles of Genetics, 2d ed., 761 (HGES)
Principles of Population Genetics, 2d ed., 761 (HGES)
Progress in Clinical and Biological Research, vol. 329: *Multipoint Mapping and Linkage Analysis Based upon Affected Pedigree Members: Genetic Analysis Workshop 6*, 1031
Tourette Syndrome and Human Behavior, 362, 363
Turner Syndrome, 595
 BrdUrd antibody method, replication patterns of fragile X, 988
- Carcinoma, medullary thyroid with parathyroid tumors, chromosome 10, 946
 Cardiac myosin heavy chain genes, cardiomyopathy, 389
 Cardiomyopathy, familial hypertrophic, CRI-L436 and CRI-L329 genes, 389
 Carrier screening
 Cystic fibrosis, 603 (E), 740 (L)
 Autosomal recessive diseases, 359 (L)
 Ethics, 580 (L)
 Cell lines, family, research, 1029 (L)
 Charcot-Marie-Tooth disease
 Type I, genetic linkage and heterogeneity, 915
 Type 1A
 Chromosome 17p11.2-p12, 680
 Gene marker, 926
 Chimerism, pigmentary anomalies, 166 (L), 167 (L)
 Chorionic villi, cytogenetic analysis, spontaneous abortion, 656
 Choroideremia, classical, deletions, 622
 Chromosomal mosaicism, pigmentary anomalies, 166 (L), 167 (L)
 Chromosome 1, markers, mapping, ovarian teratoma, 644
 Chromosome 1q25, autosomal recessive chronic granulomatous disease, 483
 Chromosome 4p16.3, α -L-iduronidase gene, 802
 Chromosome 5
 Familial adenomatous polyposis, DNA markers, 982
 Human, double-strand break-repair deficiency and gamma-ray sensitivity in XR-1 hamster variant, 459
 Chromosome 5q11-q13, schizophrenia susceptibility gene, 524
 Chromosome 5q35.1, D₁ dopamine receptor gene, 828

- Chromosome 7q11.23, autosomal recessive chronic granulomatous disease, 483
- Chromosome 10, centromere, medullary thyroid carcinoma with parathyroid tumors, 946
- Chromosome 11p13, de novo constitutional deletions, 155
- Chromosome 11q23, ataxia-telangiectasia locus, mapping, 860
- Chromosome 12q14, autosomal recessive vitamin D dependency type I, 28
- Chromosome 14, Krabbe disease, 37
- Chromosome 14q11, angiogenin gene, 973
- Chromosome 14q11-q12, CRI-L436 and CRI-L329 genes, cardiomyopathy, 389
- Chromosome 15q11q13, DNA markers, Angelman syndrome, 149
- Chromosome 17p11.2-p12, Charcot-Marie-Tooth neuropathy type 1A, 680
- Chromosome 21, Down syndrome phenotype, 236
- Chromosome 22, alleles, meningioma, 877 (L)
- Chromosome 22, losses, sporadic meningioma, 823
- Chromosome X, synaptophysin, 551
- Chromosome Xp, microsatellite probe DXS426, 935
- Chromosome Xp21, deletion mapping, Åland Island eye disease, 795
- Chromosome Xq21, DNA markers, Allan-Herndon syndrome, linkage, 454
- Chromosome Xp21.1-Xp22.3, Nance-Horan syndrome, 13
- Chromosome Xp22.1-p22.3, probes, X-linked juvenile retinoschisis, 616
- Chromosome Xp22.3, X-linked Kallman syndrome, 664
- Chromosome Xq26-Xq28, DNA markers, genetic mapping, fragile X, 395
- Chromosome Xq26.3-q27.1, X-linked albinism-deafness syndrome, 20
- Clines, multilocus, gene frequency diversity, 867
- Cloning
- α-Actinin gene, cytoskeletal, 62
 - cDNA, PCR, methylmalonic acidemia, 808
- CYP2D locus, defective oxidation of drugs, 994
- CYP2D6 mutant allele, defective oxidation of drugs, 994
- Cysteine, substitution for α1-glycine 904 in COL1A1, osteogenesis imperfecta, 670
- Cystic fibrosis
- Carrier screening, 359 (L), 603 (E), 740 (L)
 - Ethics, 580 (L)
 - Gene, benign missense variations, 611
 - Mutations, 606
 - Population screening, 164 (L)
 - Risk analysis, slash sheet, 1024 (L)
- Cystic Fibrosis Genetic Analysis Consortium, ΔF508 mutation, survey, 354 (L)
- Cytogenetic analysis
- Direct-preparation method, spontaneous abortion, 656
 - Isochromosome 18p syndrome, 493
 - Ovarian teratoma, 635
- D₁ dopamine receptor gene, 5q35.1, *Eco*RI RFLP, 828
- D3S47, autosomal dominant retinitis pigmentosa, linkage, 536
- D9S5, linkage disequilibrium with Friedreich ataxia, 228
- D9S15, linkage disequilibrium with Friedreich ataxia, 228
- D10S94, multiple endocrine neoplasia type 2A, 952
- Deletion(s)
- α1-Antitrypsin coding exons, α1-antitrypsin deficiency allele, 403
 - β^s Chromosomes, Bantu haplotype, 161
 - Classical choroideremia, 622
 - De novo constitutional, chromosome 11p13, 155
 - Factor IX_{Seattle} 1, 1020
 - Mapping, Åland Island eye disease, Xp21, 795
 - Mutations causing hemophilia B, 202
 - Nucleus-driven large-scale, mitochondrial genome, 904
 - Partial, aldolase B gene in hereditary fructose intolerance, 101
 - Repeat sequence, E1α gene, PDH deficiency, 286
- ΔF508 mutation, survey, 354 (L)
- Deoxyribonuclease, serum, polymorphism, 121
- Diabetes mellitus, insulin-dependent, T-cell receptor genes, 45
- Diagnosis
- Cytogenetic, isochromosome 18p syndrome, 493
 - Prenatal
 - β-Thalassemia, 369
 - Type III glycogen storage disease, 735
- Diagnostic tests, DNA, clinical care, 583 (L)
- Dietary environment, apo A-I, 414
- Dihydropteridine reductase deficiency, threonine, 279
- Disease, autosomal recessive, carrier screening, 359 (L)
- Disequilibrium, linkage, D9S5 and D9S15 and Friedreich ataxia, 228
- DNA
- Complementary, cloning, PCR, methylmalonic acidemia, 808
 - Diagnostic tests, clinical care, 583 (L)

- Double-strand break-repair deficiency and gamma-ray sensitivity, XR-1 hamster variant, 459
- Fingerprints, cell-line individualization, 499
- Haplotype analysis, hyperphenylalaninemia, 706
- Marker(s)
 - Chromosome 15q11q13, Angelman syndrome, 149
 - D10S94, multiple endocrine neoplasia type 2A, 952
 - Polymorphic, genetic linkage map for familial adenomatous polyposis, 982
 - Xq21, Allan-Herndon syndrome, linkage, 454
 - Xq26-Xq28, genetic mapping, fragile-X locus, 395
- Mitochondrial, polymorphism and heterogeneity, 87
- Polymorphisms, de novo dup(21q) in Down syndrome, 968
- Probes near *FRAXA*, physical mapping, 187
- Typing, forensic medicine, 876 (L)
- Dopamine receptor gene D₁, 5q35.1 *EcoRI*, 828
- Double-strand break-repair deficiency, DNA, XR-1 hamster variant, 459
- Down syndrome
 - De novo dup (21q), 968
 - Detection, gestational age, 581 (L), 583 (L)
 - DNA polymorphisms, 968
 - Phenotype, chromosome 21, 236
 - Thyroid antibodies as risk factor, 727
- Drug oxidation, defective, *CYP2D* locus, 994
- dup (21) Chromosomes, de novo, Down syndrome, 968
- DXS426, microsatellite probe, X-linked retinitis pigmentosa, 935
- E1 α gene, repeat sequence deletion, pyruvate dehydrogenase deficiency, 286
- Education, medical school curriculum, genetics, 748 (HGES-E), 753 (HGES)
- Survey, 750 (HGES)
- Ehler-Danlos syndrome type IV, familial aneurysms, 112
- Endocrine neoplasia, multiple type 2A, D10S94, 952
- Enzymatic amplification, allele and genotype frequencies, HLA-DQ α , 515
- Epidemiology, genetic, hereditary tyrosinemia, 302
- Ethical/legal issues
 - Cystic fibrosis carrier screening, 580 (L)
 - Linkage analysis in Huntington disease, 1 (E)
 - Predictive testing, adult-onset disease, 4
- Ethnogeny, HLA antigen distribution, 294
- Etiology, senile systemic amyloidosis, 127
- Fabry disease, hemizygoties, α -Gal A point mutations, 784
- Facioscapulohumeral muscular dystrophy, linkage analysis, 376
- Factor VIII, gene point mutations, hemophilia A, 743 (L)
- Factor IX, germ-line mutation pattern, 835
- Factor IX_{Seattle 1}, gene deletion by recombination, 1020
- Family cell lines, research, 1029 (L)
- Fibroblast(s)
 - Cultured skin, alkaline phosphatase, 776
 - mut^o* methylmalonic acidemia, heterozygous mutations, 808
 - Normal and hypophosphatasia, alkaline phosphatase, 767
- Forensic medicine, DNA typing, 876 (L)
- Fragile-site expression, genetic determination, 196
- Fragile X, 175 (E)
 - DNA probes near, 187
 - Microdissection, 181
 - Replication patterns in heterozygous carriers, 988
 - Xq26-Xq28, DNA markers, 395
- Friedreich ataxia, genetic homogeneity and linkage disequilibrium with D9S5 and D9S15, 228
- Fructose intolerance, hereditary
 - Nonsense mutation, aldolase B gene, 562
 - Partial aldolase B gene deletions, 101
- Fumarylacetoacetate, hereditary tyrosinemia type I, mass screening, 325
- Fumarylacetoacetate hydrolase deficiency, hereditary tyrosinemia, 308
- G1934 \rightarrow A, mutant *CYP2D6* allele, defective oxidation of drugs, 994
- Gamma-ray sensitivity, XR-1 hamster variant, 459
- Gaucher disease
 - Acid β -glucosidase gene alleles, 79
 - Type III, glucocerebrosidase gene mutation, 275
- Gene-disease associations, testing, 266
- Genetic counseling, rare syndromes, 53
- Genetic epidemiology, hereditary tyrosinemia, 302
- Genetic heterogeneity, autosomal dominant retinitis pigmentosa, 536
- Genetic linkage, mapping of Krabbe disease, 37
- Genetic mosaicism, pigmentary anomalies, 166 (L), 167 (L)
- Genomic imprinting, absence in sporadic meningioma, 823
- Genotype frequency, HLA-DQ α , 515
- Germ-line mutation pattern, factor IX, 835
- Gestational age, MSAFP screening and Down syndrome detection, 581 (L), 583 (L)
- Glucocerebrosidase gene mutation, Gaucher disease type III, 275

- Glucose-6-phosphate dehydrogenase
 Andulus^{1361A}, 575
 Mediterranean, 1008, 1013
 Molecular genetics, 575
 NT 1311 polymorphisms, 1008
 Glycogen storage disease, type III, prenatal diagnosis, 735
 Glycoprotein, disulfide-bonded salivary proline-rich, *PRB3* alleles, 686
 Granulomatous disease, chronic autosomal recessive, gene assignment, 483
 Gyrate atrophy, ornithine aminotransferase locus, splicing defect, 790
- Haplotype
 Bantu, deletion in β^s chromosomes, 161
 7, phenylketonuria genes, termination mutation, 1002
- Haplotype analysis
 apo B mutation and familial defective apo B100, 712
 DNA, hyperphenylalaninemia, 706
- Hemizygotes, Fabry disease, α -Gal A point mutations, 784
- Hemochromatosis, genetic, segregaton/linkage analysis, 542
- Hemoglobin C mutation, allele-specific PCR, 1023 (L)
- Hemophilia A, clinical manifestations, FVIII point mutations, 743 (L)
- Hemophilia B, mutations, 202
- Heteroduplexes
 Formation in PCR, 169 (L)
 PCR-generated in Tay-Sachs carriers, 169 (L)
- Heterogeneity
 Acid alpha glucosidase deficiency, 73
 Charcot-Marie-Tooth disease type I, 915
 Genetic, autosomal dominant retinitis pigmentosa, 536
 mtDNA polymorphism, 87
 Phenotypic, osteogenesis imperfecta, 670
 Risk calculation, 165 (L)
- Heterozygote, compound, lipoprotein lipase deficiency, 721
- Heterozygous carriers, replication patterns of fragile X, 988
- Heterozygous mutations, *mut*^o methylmalonic acidemia fibroblasts, 808
- Hex A* mutant alleles, frequency, Tay-Sachs screening, 698
- Hexosaminidase, pseudodeficiency, 880 (L), 881 (L)
- History, genetics, 601 (E)
- HLA, segregation/linkage analysis, genetic hemochromatosis, 542
- HLA antigen distribution, ethnogeny, 286
- HLA-DQ α allele, frequency, 515
- Homogeneity, genetic, Friedreich ataxia, 228
- HRAS1 locus, minisatellite allele diversification, 854
- Huntington disease
 Linkage analysis, ethical/legal issues, 1 (E)
 Predictive testing, ethical/legal issues, 4
- Hybrids, chromosome 17-retaining, *Alu*-PCR, Charcot-Marie-Tooth disease type 1A gene marker, 926
- Hybridization, in situ, cytogenetic diagnosis, isochromosome 18p syndrome, 493
- Hyperphenylalaninemia, DNA haplotype analysis, 706
- Hypervariable repeat sequence CRI-S232, X-linked Kallman syndrome, 664
- Hypogonadism, hypogonadotropic, Xp22.3, 664
- Hypophosphatasia, fibroblasts, alkaline phosphatase, 767
- Identity-by-descent sharing, relatives, probabilities, 842
- Imprinting, genomic, absence in sporadic meningioma, 823
- Inheritance, RNA splicing mutation in COL3A1, 112
- Insertion mutation, Tay-Sachs carriers, PCR-generated heteroduplexes, 169 (L)
- Interference coefficients, multilocus linkage analysis, 255
- Isochromosomes, de novo dup(21q) in Down syndrome, 968
- Isochromosome 18p syndrome, cytogenetic diagnosis, 493
- Isoenzyme, tissue nonspecific, alkaline phosphatase, 767
- Isozymes, pattern similarities in serum, urine, and organs, 121
- Kallman syndrome, X-linked, X-22.3, 664
- Krabbe disease, mapping by genetic linkage, 37
- Landouzy-Déjérine muscular dystrophy, linkage analysis, 376
- Leber hereditary optic neuropathy, absence of mitochondrial ND4 gene mutation, 629
- Leber hereditary optic neuroretinopathy, segregation of mitochondrial genomes, 95
- Legal/ethical issues
 Linkage analysis in Huntington disease, 1 (E)
 Predictive testing, adult-onset disease, 4
- Linkage
 Affected sib pairs, 45
 Allan-Herndon syndrome and Xq21 DNA markers, 454
 α -Actinin gene and β -spectrin gene, 62

- Autosomal dominant retinitis pigmentosa and D3S47, 536
- Genetic
- Detection by multivariate method, 247
 - Type I Charcot-Marie-Tooth disease, 915
- Genetic map
- DNA markers for familial adenomatous polyposis, 982
 - Krabbe disease, 37
- Microsatellite probe DXS426 and X-linked retinitis pigmentosa, 935
- Osteogenesis imperfecta and COL1A1 and COL1A2, 592 (L), 593 (L)
- X-linked
- Juvenile retinoschisis and Xp22.1-p22.3 probes, 616
 - Kallman syndrome and CRI-S232, 664
- Linkage analysis
- Dominant locus for quantitative trait, lod score method, 218
 - Facioscapulohumeral muscular dystrophy, 376
 - Genetic hemochromatosis, 542
 - Huntington disease, ethical/legal issues, 1 (E)
 - Localization of Nance-Horan syndrome, 13
 - Mapping autosomal recessive vitamin D dependency type I, 28
 - Multilocus, recombination fractions and interference coefficients, 255
- Linkage disequilibrium, D9S5 and D9S15 and Friedrich ataxia, 228
- Lipoprotein, apo AI-CIII-AIV gene cluster and apo B gene loci, 429
- Lipoprotein lipase
- Deficiency
 - Compound heterozygote, 721
 - Nonsense mutation in LPL gene, 107
- Gene
- AG→AA transition, 721
 - Nonsense mutation, 107
- Lipoprotein phenotype, detection of genetic linkage by multivariate method, 247
- Map/mapping
- Ataxia-telangiectasia locus, 11q23, 860
 - Autosomal recessive vitamin D dependency type I, 28
 - Chromosome 1 markers, ovarian teratoma, 644
 - Deletion, Åland Island eye disease, Xp21, 795
 - Genetic, Xq26-Xq28 DNA markers, fragile-X locus, 395
 - Genetic linkage
 - DNA markers for familial adenomatous polyposis, 982
 - Krabbe disease, 37
- Physical, DNA probes near FRAXA, 187
- X-linked albinism-deafness syndrome, 20
- Marker(s)
- Charcot-Marie-Tooth disease type 1A gene, 926
 - Chromosome 1, gene-centromere mapping, ovarian teratoma, 644
- DNA
- Chromosome 15q11q13, Angelman syndrome, 149
 - D10S94, multiple endocrine neoplasia type 2A, 952
 - Polymorphic, genetic linkage map for familial adenomatous polyposis, 982
 - Xq21, Allan-Herndon syndrome, linkage, 454
 - Xq26-Xq28, genetic mapping, fragile-X locus, 395
 - Loci D9S5 and D9S15, linkage disequilibrium with Friedrich ataxia, 228
 - Nonsyntenic loci COL1A1 and COL1A2, OI linkage, 593 (L), 593 (L)
- Maternal serum alpha-fetoprotein
- Policy statement update, remarks on criticism, 740 (L)
- Screening
- Gestational age, 581 (L), 583 (L)
 - State health agencies, 899
 - State activities, 896 (E)
- Medical school curriculum, genetics, 748 (HGES-E), 753 (HGES)
- Survey, 750 (HGES)
- Medullary thyroid carcinoma with parathyroid tumors, chromosome 10, 946
- Membership survey results, American Society of Human Genetics, 345
- Menarche, age as fitness trait, 137
- Meningioma
- Chromosome 22 alleles, 877 (L)
 - Sporadic, chromosome 22 losses, 823
- Methylmalonic acidemia, *mut*^o, fibroblast heterozygous mutations, 808
- Microsatellite probe DXS426, X-linked retinitis pigmentosa, 935
- Minisatellite allele diversification, HRAS1 locus, 854
- Missense variations, benign, cystic fibrosis gene, 611
- Mitochondrial DNA, polymorphism and heterogeneity, 87
- Mitochondrial genome(s)
- Nucleus-driven large-scale deletions, 904
 - Segregation in Leber hereditary optic neuroretinopathy, 95
- Mitochondrial ND4 gene, mutation, absence in Leber hereditary neuropathy, 629
- Monte Carlo comparison, ordering multiple loci, 470
- Mosaicism, chromosomal and genetic, pigmentary

- anomalies, 166 (L), 167 (L)
- Multilocus clines, gene frequency diversity, 867
- Multilocus linkage analysis, recombination fractions and interference coefficients, 255
- Multipoint analysis, quantitative variation, 957
- Muscular dystrophy, facioscapulohumeral, linkage analysis, 376
- Mutation(s)
 - apo B, haplotype analysis, 712
 - Cystein for α 1-glycine 904 in COL1A1, osteogenesis imperfecta, 670
 - Cystic fibrosis, 606
 - Δ F508, survey, 354 (L)
 - Familial defective apo B100, 712
 - Fragile X, DNA probes near, 187
 - Germ line, factor IX, 835
 - Glucose-6-phosphate dehydrogenase Mediterranean, 1008
 - Hemoglobin C, allele-specific PCR, 1023 (L)
 - Hemophilia B, 202
 - Heterozygous, *mut*^o methylmalonic acidemia fibroblasts, 808
 - Insertion, Tay-Sachs carriers, PCR-generated heteroduplexes, 169 (L)
 - Medullary thyroid carcinoma with parathyroid tumors, chromosome 10, 946
 - Mitochondrial ND4 gene, absence in Leber hereditary neuropathy, 629
 - Nonsense
 - Aldolase B gene, hereditary fructose intolerance, 562
 - Lipoprotein lipase gene, 107
 - Point
 - α -Galactosidase A gene, Fabry disease, 784
 - β -Hexosaminidase α subunit gene, infantile Tay-Sachs disease, 568
 - FVIII gene, hemophilia A, 743 (L)
 - RNA splicing, inheritance, 112
 - Termination, haplotype 7 phenylketonuria genes, 1002
- mut*^o methylmalonic acidemia, fibroblast heterozygous mutations, 808
- Nance-Horan syndrome, localization by linkage analysis, 13
- Neoplasia, multiple endocrine type 2A, D10S94, 952
- Neuropathy
 - Charcot-Marie-Tooth type 1A, chromosome 17p11.2-p12, 680
 - Leber hereditary optic, absence of mitochondrial ND4 gene mutation, 629
 - Motor and sensory, type I hereditary, genetic linkage and heterogeneity, 915
- Neuroretinopathy, Leber hereditary optic, segregation of mitochondrial genomes, 95
- New Developments in Biotechnology*, critique of book review, 1028 (L)
- Nondisjunction, origin, ovarian teratoma, 644
- Nonsense mutation
 - Aldolase B gene, hereditary fructose intolerance, 562
 - Lipoprotein lipase gene, 107
- Nonsynthetic marker loci, CO1A1 and COL1A2, osteogenesis imperfecta linkage, 592 (L), 593 (L)
- NT 1311 polymorphism, glucose-6-phosphate dehydrogenase, 1008
- Oligonucleotide probes, allele and genotype frequencies, HLA-DQ α , 515
- Ornithine aminotransferase locus, splicing defect, gyrate atrophy, 790
- Osteogenesis imperfecta, phenotypic heterogeneity, 670
- Ovarian teratoma, cytogenetic analysis and mechanism origin, 635, 644
- Pathology, visceral, hereditary tyrosinemia type I, 317
- Pedigree, linkage, resampling method for confidence interval, 53
- Phenotype
 - Down syndrome, 236
 - Lipoprotein, detection of genetic linkage by multivariate method, 247
- Phenylketonuria, haplotype 7 genes, termination mutation, 1002
- Phosphoethanolamine, alkaline phosphatase, 767
- Pigmentary anomalies, mosaicism and chimerism, 166 (L), 167 (L)
- Point mutation(s)
 - α -Galactosidase A gene, Fabry disease, 784
 - β -Hexosaminidase α subunit gene, infantile Tay-Sachs disease, 568
 - FVIII gene, hemophilia A, 743 (L)
- Policy statement update, MSAFP, remarks on criticism, 740 (L)
- Polymerase chain reaction
 - Allele-specific, hemoglobin C mutation, 1027
 - Alu*, chromosome 17-retaining hybrids, Charcot-Marie-Tooth disease type 1A gene marker, 926
 - cDNA cloning, methylmalonic acidemia, 808
 - Generation of heteroduplexes from Tay-Sachs carriers, 169 (L)
 - Heteroduplex formation, 169 (L)
- Polymorphism(s)
 - DNA, de novo dup(21q) in Down syndrome, 968
 - mtDNA, heterogeneity, 87

- NT 1311, glucose-6-phosphate dehydrogenase, 1008
 Restriction-fragment-length, *EcoRI*, DRD1, 828
 Serum deoxyribonuclease, 121
 Polyposis, familial adenomatous, polymorphic DNA markers, 982
 Population screening, cystic fibrosis, 164 (L)
 PRB3 locus, alleles, proline-rich gl 8, 686
 Predictive testing, adult-onset disease, 1 (E)
 Ethical/legal issues, 4
 Pregnancy wastage, cytogenetic analysis, spontaneous abortion, 656
 Prenatal diagnosis
 β -Thalassemia, 369
 Glycogen storage disease type III, 735
 Procollagen gene, RNA splicing mutation, inheritance, 112
 Procollagen type I gene, mutation, cysteine for α 1-glycine, osteogenesis imperfecta, 670
 Proline \rightarrow histidine substitution, rhodopsin, absence in autosomal dominant retinitis pigmentosa, 941
 Pseudodeficiency, hexosaminidase, 880 (L), 881 (L)
 Pseudohypophosphatasia, alkaline phosphatase, 776
 Pyridoxal-5'-phosphate ectophosphatase, alkaline phosphatase, 767
 Pyruvate dehydrogenase deficiency, E1 α gene, repeat sequence deletion, 286
 Quantitative trait, linkage analysis of dominant locus, 218
 Quantitative variation, multipoint analysis, 957
 Recombination, gene deletion in factor IX_{Seattle1}, 1020
 Recombination fraction, multilocus linkage analysis, 255
 Recurrence risks, live births and "stillbirths," 741 (L), 742 (L)
 Replication patterns, fragile X in heterozygous carriers, 988
 Restriction-fragment-length polymorphism, *EcoRI*, DRD1, 828
 Retinitis pigmentosa
 Autosomal dominant
 Absence of rhodopsin pro \rightarrow his substitution, 941
 D3S47 linkage, 536
 X-linked, microsatellite probe DXS426, 935
 Retinoschisis, X-linked juvenile, Xp22.1-p22.3 probes, linkage, 616
 Rhodopsin, proline \rightarrow histidine substitution, absence in ADRP, 941
 Risk
 Calculation under heterogeneity, 165 (L)
 Genetic, support intervals, 166 (L)
 Risk analysis, cystic fibrosis, slash sheet, 1024 (L)
 Risk factor, thyroid antibodies, Down syndrome and other trisomies, 727
 RNA splicing mutation, COL3A1, inheritance, 112
 Salivary proline-rich glycoprotein, disulfide bonded, PRB3 alleles, 686
 Schizophrenia, susceptibility gene, chromosome 5q11-q13, 524
 Screening
 Carrier
 Cystic fibrosis, 603 (E), 740 (L)
 Autosomal recessive disease, 359 (L)
 Ethics, 580 (L)
 Hereditary tyrosinemia type I, fumarylacetoacetate, 325
 Maternal serum alpha-fetoprotein
 Gestational age, 581 (L), 583 (L)
 State health agencies, 899
 Patent of test, 353 (L)
 Population, cystic fibrosis, 164 (L)
 Tay-Sachs disease, *Hex A* mutant alleles frequency, 698
 Segregation, mitochondrial genomes in Leber hereditary optic neuroretinopathy, 95
 Segregation analysis, genetic hemochromatosis, 542
 Sex ratio
 Progenitive, sperm cells, 351 (L)
 Sperm cells, 349 (L)
 Sib pairs, affected, linkage, 45
 Slash sheet, risk analysis in cystic fibrosis, 1024 (L)
 Social Issues Committee report, American Society of Human Genetics, 343
 Sperm cells
 Progenitive sex ratio, 351 (L)
 Sex ratio, 349 (L)
 Splicing defect, ornithine aminotransferase locus, gyrate atrophy, 790
 Splicing mutation, RNA, inheritance, 112
 Support intervals, genetic risk, 166 (L)
 Synaptophysin, gene structure/assignment, 551
 T-cell receptor α/β locus, 14q11, 973
 T-cell receptor genes, insulin-dependent diabetes mellitus, 45
 Tay-Sachs disease
 Infantile
 β -Hexosaminidase α subunit gene, point mutation, 568
 Mutant alleles, 815
 PCR-generated heteroduplexes, 169 (L)
 Screening, *Hex A* mutant alleles frequency, 698

- Teratoma, ovarian, cytogenetic analysis and mechanism of origin, 635, 644
- Termination mutation, haplotype 7 phenylketonuria genes, 1002
- Threonine, dihydropteridine reductase deficiency, 279
- Thyroid antibodies, Down syndrome and other trisomies, 727
- Thyroid carcinoma, medullary with parathyroid tumors, chromosome 10, 946
- Transient gene expression, acid alpha glucosidase allele, 440
- Transition(s)
- Lipoprotein lipase deficiency, 721
 - Mutations causing hemophilia B, 202
- Translocations, de novo dup(21q) in Down syndrome, 968
- Transplantation, liver, hereditary tyrosinemia, 338
- Transthyretin variant, homozygous, senile systemic amyloidosis, 127
- Transversions, mutations causing hemophilia B, 202
- Trisomy, thyroid antibodies as risk factors, 727
- Tumors, parathyroid, medullary thyroid carcinoma, 946
- Twins
- Age at menarche as fitness trait, 137
 - Appendectomy, 590 (L)
- Tyrosinemia, hereditary
- Fumarylacetoacetate hydrolase deficiency, 308
 - Genetic epidemiology, 302
 - Liver transplantation, 338
- Type I
- Homogentisic acid, oral loading, 329
 - Screening, fumarylacetoacetate, 325
 - Visceral pathology, 317
- Vitamin D dependency type I, autosomal recessive, mapping, 28
- X-linked albinism-deafness syndrome, mapping, 20
- X-linked juvenile retinoschisis, Xp22.1-p22.3 probes, linkage, 616
- X-linked Kallman syndrome, Xp22.3, 664
- X-linked retinitis pigmentosa, microsatellite probe DXS426, 935
- Xr-1 hamster variant, DNA double-strand break-repair deficiency and gamma ray sensitivity, 459

